

NIEHS-supported researchers map epigenome of more than 100 tissue and cell types

By Joe Balintfy

NIEHS grantees, supported by the National Institutes of Health (NIH) Common Fund's [Roadmap Epigenomics Program](http://commonfund.nih.gov/epigenomics/index)

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, have mapped the epigenomes of more than 100 types of cells and tissues, providing new insight into which parts of the genome are used to make a particular type of cell. The epigenome is part of the machinery that helps direct how genes are turned off and on in different types of cells.

Much like mapping the human genome laid the foundations for understanding the genetic basis of human health, these new maps of the human epigenome may further unravel the complex links between DNA and disease.

"This represents a major advance in the ongoing effort to understand how the 3 billion letters of an individual's DNA instruction book are able to instruct vastly different molecular activities, depending on the cellular context," said NIH Director Francis Collins, M.D., Ph.D. "This outpouring of data-rich publications, produced by a remarkable team of creative scientists, provides powerful momentum for the rapidly growing field of epigenomics."

Researchers from the Roadmap Epigenomics Program published Feb. 19 a description of the epigenome maps in the journal [Nature](http://www.ncbi.nlm.nih.gov/pubmed/25693563)

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. More than 20 additional papers, published in *Nature* and *Nature*-associated journals, show how these maps can be used to study human biology.

"What the Roadmap Epigenomics Program has delivered is a way to look at the human genome in its living, breathing nature from cell type to cell type," said senior author Manolis Kellis, Ph.D., professor of computer science at the Massachusetts Institute of Technology, and an NIEHS grantee.

Linked Video

[Watch "Epigenome: The Symphony in your Cells," a Nature video that uses music to explain the epigenome.](#)

Understanding epigenomics

Almost all human cells have identical genomes that contain instructions on how to make the many different cells and tissues in the body. During the development of different types of cells, regulatory proteins turn genes on and off and, in doing so, establish a layer of chemical signatures that make up the epigenome of each cell. In the Roadmap Epigenomics Program, researchers compared these epigenomic signatures and established their differences across a variety of cell types. The resulting information can help us understand how changes to the genome and epigenome can lead to conditions such as Alzheimer's disease, cancer, asthma, and fetal growth abnormalities.

"This is the most comprehensive catalog of epigenomic data from primary human cells and tissues to date," said [Lisa Chadwick, Ph.D.](#), project team leader and program director at NIEHS. "This coordinated effort, along with uniform data processing, makes it much easier for researchers to make direct comparisons across the entire data set."

The data, available to the biomedical research community, can be found at the [National Center for Biotechnology Information website](http://www.ncbi.nlm.nih.gov/).

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The value of epigenomic data



"Epigenomics represents the next phase in our understanding of genetic regulation of health and disease," Collins said. (Photo courtesy of NIH)



Chadwick was interviewed about the epigenome papers by major media outlets including the Boston Globe, PBS Newshour, and Reuters. (Photo courtesy of Steve McCaw)

Researchers can now take data from different cell types and directly compare them. “Today, sequencing the human genome can be done rapidly and cheaply, but interpreting the genome remains a challenge,” said NIEHS grantee [Bing Ren, Ph.D.](#) (<http://igm.ucsd.edu/faculty/profiles/bing-ren.shtml>)

, professor of cellular and molecular medicine at the University of California, San Diego, and co-author of the Nature paper and several of the associated papers. “These 111 reference epigenome maps are essentially a vocabulary book that helps us decipher each DNA segment in distinct cell and tissue types. These maps are like snapshots of the human genome in action.”

“Researchers from the 88 projects supported by the program, including those from this recent series of papers, have propelled the development of new epigenomic technologies,” said [John Satterlee, Ph.D.](#) (<http://www.drugabuse.gov/about-nida/organization/divisions/division-basic-neuroscience-behavioral-research-dbnbr/genetics-molecular-neurobiology-research-branch-gmnr/b>)

, co-coordinator of the Roadmap Epigenomics Program, and program director at the National Institute on Drug Abuse (NIDA), part of NIH. Satterlee added that the work of this program has served as a foundation for continued exploration of the human epigenome through the [International Human Epigenome Consortium](#) (<http://www.ihc-epigenomes.org/>)

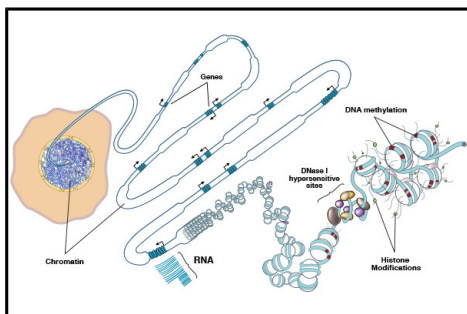
NIDA, NIEHS, and the National Institute on Deafness and Other Communication Disorders are co-administrators of the NIH Common Fund’s Epigenomics Program.

Citation: [Roadmap Epigenomics Consortium](#)

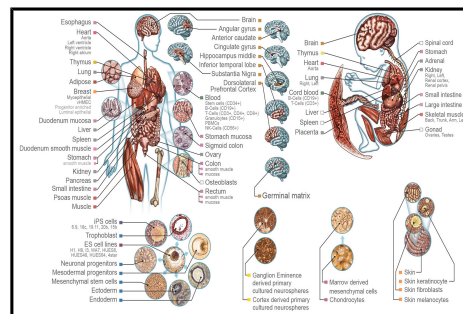
(<http://www.ncbi.nlm.nih.gov/pubmed/25693563>)

. 2015. Integrative analysis of 111 reference human epigenomes. Nature 518(7539):317-330.

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An epigenomic signature can be made on the genome in two ways, both of which play a role in turning genes off or on. The first occurs when chemical tags called methyl groups are attached to a DNA molecule directly (DNA methylation). The second occurs when a variety of chemical tags attach to the tails of histone proteins that package DNA (histone modifications).



Reference epigenomes are available for more than 100 cell and tissue types. (Photo courtesy of Nature and Roadmap Epigenomics Consortium)

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